2005 Contents

Volume 69 Number 1 January 2005

- 1 Phenotype Severity and Genetic Variation at the Disease Locus: An Investigation of Nail Dysplasia in the Nail Patella Syndrome
 - J. A. Dunston, S. Lin, J. W. Park, M. Malbroux and I. McIntosh
- 9 Double Heterozygosity with Mutations Involving both the GJB2 and GJB6 Genes is a Possible, but very Rare, Cause of Congenital Deafness in the Czech Population
 P. Seeman, O. Bendová, D. Rašková, M. Malíková, D. Groh and Z. Kabelka
- 15 Comprehensive Cystic Fibrosis Mutation Epidemiology and Haplotype Characterization in a Southern Italian Population
 G. Castaldo, A. Polizzi, R. Tomaiuolo, C. Cazeneuve, E. Girodon, T. Santostasi, D. Salvatore, V. Raia, N. Rigillo, M. Goossens and F. Salvatore
- 25 The Contribution of Genetic and Environmental Factors to Quantitative Variability of Erythrocyte Membrane Proteins in Primary Hypotension
 V. P. Ivanov, A. V. Polonikov and M. A. Solodilova
- 36 β₂-Adrenergic Receptor Gene Variations Associated with Stage-2 Hypertension in Northern Han Chinese D. Ge, J. Huang, J. He, B. Li, X. Duan, R. Chen and D. Gu
- 45 Linkage Analysis of Chromosome 1 with Essential Hypertension and Blood Pressure Quantitative Traits in Chinese Families
 D. Ge, J. Huang, W. Yang, J. Zhao, Y. Shen, B. Qiang and D. Gu
- 55 Genetic Heterogeneity of Beta Thalassemia in Lebanon Reflects Historic and Recent Population Migration N. J. Makhoul, R. S. Wells, H. Kaspar, H. Shbaklo, A. Taher, N. Chakar and P. A. Zalloua
- 67 Mitochondrial DNA Diversity in Indigenous Populations of the Southern Extent of Siberia, and the Origins of Native American Haplogroups
 E. B. Starikovskaya, R. I. Sukernik, O. A. Derbeneva, N. V. Volodko, E. Ruiz-Pesini, A. Torroni, M. D. Brown, M. T. Lott, S. H. Hosseini, K. Huoponen and D. C. Wallace
- 90 Genetic Association Studies in Complex Disease: Disentangling Additional Predisposing Loci from Associated Neutral Loci Using a Constrained - Permutation Approach G.T. Spijker, I.M. Nolte, R.C. Jansen and G.J. Te Meerman
- 102 Fine Mapping Functional Sites or Regions from Case-Control Data Using Haplotypes of Multiple Linked SNPs
 R. Cheng, J. Z. Ma, R. C. Elston and M. D. Li
- 113 Linkage Analysis of Affected Sib Pairs Allowing for Parent-of-Origin Effects C.-C. Wu, S. Shete and C. I. Amos
- 127 The use of Allelic Expression Differences to Ascertain Functional Polymorphisms Acting in cis: Analysis of MMP1 Transcripts in Normal Lung Tissue
 J. Heighway, N. L. Bowers, S. Smith, D. C. Betticher and M. F. Santibáëz Koref
- 134 Corrigendum

Volume 69 Number 2 March 2005

- Analysis of Y-chromosome Variability and its Comparison with mtDNA Variability Reveals Different Demographic Histories Between Islands in the Azores Archipelago (Portugal)
 R. Montiel, C. Bettencourt, C. Silva, C. Santos, M. J. Prata and M. Lima
- The Y-chromosomal Heritage of the Azores Islands Population
 P. R. Pacheco, C. C. Branco, R. Cabral, S. Costa, A. L. Araújo, B. R. Peixoto, P. Mendonça and L. Mota-Vieira
- Linkage Analysis of Plasma ApoE in Three Ethnic Groups: Multiple Genes with Context-Dependent Effects
 K. L. E. Klos, S. L. R. Kardia, J. E. Hixson, S. T. Turner, C. Hanis, E. Boerwinkle and C. F. Sing
- 168 Haplotype Effects on Human Survival: Logistic Regression Models Applied to Unphased Genotype Data Q. Tan, L. Christiansen, L. Bathum, J. H. Zhao, W. Vach, J. W. Vaupel, K. Christensen and T. A. Kruse

- Generating Genetic Risk Scores from Intermediate Phenotypes for Use in Association Studies of Clinically Significant Endpoints
 B. D. Horne, J. L. Anderson, J. F. Carlquist, J. B. Muhlestein, D. G. Renlund, T. L. Bair, R. R. Pearson and N. J. Camp
- 187 High Resolution T^2 Association Tests of Complex Diseases Based on Family Data Ruzong Fan, Michael Knapp, Marthias Wjst, Caixia Zhao and Momiao Xiong
- 209 The Effect of SNP Marker Density on the Efficacy of Haplotype Tagging SNPs a Warning Mark M. Iles
- 216 Spinal Muscular Atrophy Carrier Screening by Multiplex Polymerase Chain Reaction using Dried Blood Spot on Filter Paper R. Majumdar, Z. Rehana, M. Al Jumah and N. Fetaini
- 222 dHPLC Screening of the NSD1 gene Identifies Nine Novel Mutations Summary of the first 100 Sotos Syndrome Mutations
 Linea Melchior, Marianne Schwartz and Morten Duno
- SNP Haplotypes in the Angiotensin I-Converting Enzyme (ACE) Gene: Analysis of Nigerian Family Data Using Gamete Competition Models
 C. A. McKenzie, J. S. Sinsheimer, A. A. Adeyemo, R. D. Cox, L. Southam, A. Hugill, N. Bouzekri, M. Lathrop, T. E. Forrester, R. S. Cooper and R. Ward
- 233 A Method for Pooling Alleles from Different Genotyping Experiments Y. S. Aulchenko, A. M. Bertoli-Avella and C. M. van Duijn

Volume 69 Number 3 May 2005

- 239 Nucleotide Sequence Analyses of Human Complement 6 (C6) Gene Suggest Balancing Selection M. Soejima, H. Tachida, M. Tsuneoka, O. Takenaka, H. Kimura and Y. Koda
- Analysis of Dystrophin Gene Deletions Indicates that the Hinge III Region of the Protein Correlates with Disease Severity
 A. Carsana, G. Frisso, M. R. Tremolaterra, R. Lanzillo, D. F. Vitale, L. Santoro and F. Salvatore
- 260 Simultaneous Selection of the Wild-type Genotypes of the G894T and 4B/4A Polymorphisms of NOS3 Associate with High-altitude Adaptation
 A. Ahsan, T. Norboo, M. A. Baig and M. A. Q. Pasha
- 268 Wilson Disease: High Prevalence in a Mountaineous Area of Crete G. V. Z. Dedoussis, J. Genschel, T. E. Sialvera, B. Bochow, N. Manolaki, Y. Manios, E. Tsafantakis and H. Schmidt
- Ethiopia: between Sub-Saharan Africa and Western Eurasia
 A. Lovell, C. Moreau, V. Yotova, F. Xiao, S. Bourgeois, D. Gehl, J. Bertranpetit, E. Schurr and D. Labuda
- 288 The Effect of Genetic Drift in a Young Genetically Isolated Population L. M. Pardo, I. MacKay, B. Oostra, C. M. van Duijn and Y. S. Aulchenko
- 296 Power and Related Statistical Properties of Conditional Likelihood Score Tests for Association Studies in Nuclear Families with Parental Genotypes
 Z. Li, J. L. Gastwirth and M. H. Gail

Short Communications

- 315 A Dominant form of Congenital Stationary Night Blindness (adCSNB) in a Large Chinese Family X. Liu, S. Zhuang, S. Hu, F. Zhang, B. Lin, X. Li, D. Xu and S.-H. Chen
- 323 Application of the Stepwise Focusing Method to Optimize the Cost-effectiveness of Genome-wide Association Studies with Limited Research Budgets for Genotyping and Phenotyping J. Ohashi and A. G. Clark
- 329 Integrating Case-control and TDT Studies G. R. Kazeem and M. Farrall
- 337 Erratum
- 345 Erratum

Announcement

347 CEPH Genotype Database Version 10 Released

Volume 69 Number 4 July 2005

- 349 Nail Patella Syndrome Revisited: 50 Years After Linkage I. McIntosh, J. A. Dunston, L. Liu, J. E. Hoover-Fong and E. Sweeney
- Polymorphic Alu Insertions and their Associations with MHC Class I Alleles and Haplotypes in the Northeastern Thais
 D. S. Dunn, A. V. Romphruk, C. Leelayuwat, M. Bellgard and J. K. Kulski
- 373 Score Statistic to Test for Genetic Correlation for Proband-Family Design R. El Galta, C. M. Van Duijn, J. C. Van Houwelingen and J. J. Houwing-Duistermaat
- 382 HLA-DQA1, -DQB1 Polymorphism and Genetic Susceptibility to Idiopathic Dilated Cardiomyopathy in Hans of Northern China
 W. Liu, W. M. Li and N. L. Sun
- Worldwide Genetic Variation at the 3'-UTR Region of the LDLR Gene: Possible Influence of Natural Selection
 N. J. R. Fagundes, F. M. Salzano, M. A. Batzer, P. L. Deininger and S. L. Bonatto
- 401 MtDNA and Y-chromosome Variation in Kurdish Groups I. Nasidze, D. Quinque, M. Ozturk, N. Bendukidze and M. Stoneking
- 413 Genetic Polymorphisms and Haplotypes of the Human Cardiac Sodium Channel α Subunit Gene (SCN5A) in Japanese and their Association with Arrhythmia K. Maekawa, Y. Saito, S. Ozawa, S. Adachi-Akahane, M. Kawamoto, K. Komamura, W. Shimizu, K. Ueno, S. Kamakura, N. Kamatani, M. Kitakaze and J. Sawada
- 429 Family-Based Association Tests for Different Family Structures Using Pooled DNA G. Zou and H. Zhao
- 443 Y-chromosome Lineages from Portugal, Madeira and Açores Record Elements of Sephardim and Berber Ancestry
 R. Gonçalves, A. Freitas, M. Branco, A. Rosa, A. T. Fernandes, L. A. Zhivotovsky, P. A. Underhill, T. Kivisild and A. Brehm
- 455 A Haplotype Similarity Based Transmission/Disequilibrium Test under Founder Heterogeneity K. Yu, S. Zhang, I. Borecki, A. Kraja, C. Xiong, R. Myers and M. Province
- 468 G Protein β3 Subunit Gene Variants and Essential Hypertension in the Northern Chinese Han Population B. Li, D. Ge, Y. Wang, W. Zhao, X. Zhou, D. Gu and R. Chen
- 474 Multipoint Linkage Disequilibrium Mapping Using Multilocus Allele Frequency Data T. Johnson
- 498 Erratum
- 499 Errata

Volume 69 Number 5 September 2005

- 501 Molecular Genetic Analysis of SLC3A1 and SLC7A9 Genes in Czech and Slovak Cystinuric Patients Z. Škopková, E. Hrabincová, S. Štástná, L. Kozák and T. Adam
- Evidence by Expression Analysis of Candidate Genes for Congenital Heart Defects in the NF1 Microdeletion Interval
 M. Venturin, A. Bentivegna, R. Moroni, L. Larizza and P. Riva
- 517 Loci Contributing to Adult Height and Body Mass Index in African American Families Ascertained for
 - Type 2 Diabetes

 M.M. Sale, B. I. Freedman, P. J. Hicks, A. H. Williams, C. D. Langefeld, C. J. Gallagher, D. W. Bowden and
 S. S. Rich
- 528 Evidence of a Common Founder for SCA12 in the Indian Population S. Bahl, K. Virdi, U. Mittal, M. P. Sachdeva, A. K. Kalla, S. E. Holmes, E. O'Hearn, R. L. Margolis, S. Jain, A. K. Srivastava and M. Mukerji
- 535 The Genetics of the Pre-Roman Iberian Peninsula: A mtDNA Study of Ancient Iberians
 M. L. Sampietro, D. Caramelli, O. Lao, F. Calafell, D. Comas, M. Lari, B. Agustí, J. Bertranpetit and C. Lalueza-Fox
- 549 The Influence of Past Endogamy and Consanguinity on Genetic Disorders in Northern Sweden A. H. Bittles and I. Egerbladh

- 559 A Comparison of Case-Control and Family-Based Association Methods: The Example of Sickle-Cell and Malaria
 - H. Ackerman, S. Usen, M. Jallow, F. Sisay-Joof, M. Pinder and D. P. Kwiatkowski
- 566 Design of Case-controls Studies with Unscreened Controls V. Moskvina, P. Holmans, K. M. Schmidt and N. Craddock
- 577 Using Tree-Based Recursive Partitioning Methods to Group Haplotypes for Increased Power in Association Studies
 - K. Yu, J. Xu, DC Rao and M. Province
- 590 Fishing for Pleiotropic QTLs in a Polygenic Sea L. E. Bauman, L. Almasy, J. Blangero, R. Duggirala, J. S. Sinsheimer and K. Lange

Volume 69 Number 6 November 2005

- The Significant Increase in Cardiovascular Disease Risk in APOE ε4 Carriers is Evident Only in Men Who Smoke: Potential Relationship Between Reduced Antioxidant Status and ApoE
 P. J. Talmud, J. W. Stephens, E. Hawe, S. Demissie, L. A. Cupples, S. J. Hurel, S. E. Humphries and J. M. Ordovas
- 623 Association of Common CRP Gene Variants with CRP Levels and Cardiovascular Events D. T. Miller, R. Y. L. Zee, J. Suk Danik, P. Kozlowski, D. I. Chasman, R. Lazarus, N. R. Cook, P. M. Ridker and D. J. Kwiatkowski
- 639 The Association of Mitochondrial DNA 5178 C > A Polymorphism with Plasma Lipid Levels Among Three Ethnic Groups
 S. Lal, M. Madhavan and C. K. Heng
- 645 Possible Association of the Human KCNE1 (minK) Gene and QT Interval in Healthy Subjects: Evidence from Association and Linkage Analyses in Israeli Families
 Y. Friedlander, M. Vatta, N. Sotoodelnia, R. Sinnreich, H. Li, O. Manor, J. A. Towbin, D. S. Siscovick and J. D. Kark
- 657 ATM Gene Founder Haplotypes and Associated Mutations in Polish Families with Ataxia-Telangiectasia M. Mitui, E. Bernatowska, B. Pietrucha, J. Piotrowska-Jastrzebska, L. Eng, S. Nahas, S. Teraoka, G. Sholty, A. Purayidom, P. Concannon and R. A. Gatti
- 665 Temporal Mitochondrial DNA Variation in the Basque Country: Influence of Post-Neolithic Events
 A. Alzualde, N. Izagirre, S. Alonso, A. Alonso and C. de la Rúa
- Diversity and Divergence Among the Tribal Populations of India W.S. Watkins, B. V. R. Prasad, J. M. Naidu, B. B. Rao, B. A. Bhanu, B. Ramachandran, P. K. Das, P. B. Gai, P. C. Reddy, P. G. Reddy, M. Sethuraman, M. J. Bamshad and L. B. Jorde
- Assessment of the Role of Genetic Polymorphism in Venous Thrombosis Through Artificial Neural Networks
 S. Penco, E. Grossi, S. Cheng, M. Intraligi, G. Maurelli, M. C. Patrosso, A. Marocchi and M. Buscema
- 707 Interaction Between Two Quantitative Trait Loci Affects Fetal Haemoglobin Expression C. Garner, S. Menzel, C. Martin, N. Silver, S. Best, T. D. Spector and S. L. Thein
- 715 Tests of Association Between Quantitative Traits and Haplotypes In A Reduced-Dimensional Space Q. Sha, J. Dong, R. Jiang and S. Zhang
- 733 SNP Selection for Association Studies: Maximizing Power across SNP Choice and Study Size F. Pardi, C. M. Lewis and J. C. Whittaker
- 747 Multiple Testing in the Context of Haplotype Analysis Revisited: Application to Case-Control Data T. Becker, S. Cichon, E. Jönson and M. Knapp

Short Communication

- The Peopling of Modern Bosnia-Herzegovina: Y-chromosome Haplogroups in the Three Main Ethnic Groups
 D. Marjanovic, S. Fornarino, S. Montagna, D. Primorac, R. Hadziselimovic, S. Vidovic, N. Pojskic, V. Battaglia, A. Achilli, K. Drobnic, S. Andjelinovic, A. Torroni, A. S. Santachiara-Benerecetti and O. Semino
- 764 33rd European Mathematical Genetics Meeting, EMGM 2005, Le Kremlin-Bicêtre, France, April 1st–2nd 2005

